Usher’s Syndrome: The Invisible Community

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Table of Contents

Abstract .................................................................................................................................................. 3

Introduction ............................................................................................................................................... 5

The Journey: Light to Darkness ........................................................................................................... 6

Usher's Syndrome: What is it? .............................................................................................................. 20

Transition from Denial to Acceptance ................................................................................................. 27

Works Cited ........................................................................................................................................... 29
Abstract

Mostly, the ability to use all five senses is taken for granted. Who truly is aware of the privilege to be able to see the bright, shining sun in the vibrant, blue sky; to feel the brisk, cool breeze gently blow across their skin; to hear the sounds of people’s shoes hitting the pavement while speaking high and low of their day; to taste exquisite cuisines from local to international origins; or to smell the wood burning in a fireplace on a crisp, Autumn evening. Growing up, some of my senses were not as strong. In July of 2011, Usher’s Syndrome branded two of my senses- hearing and vision. The diagnosis of this condition marked a day that will be changed forever. This condition is rare and unknown to many people. Usher’s Syndrome is a genetic condition that causes deafness and blindness. Through my research, this thesis will educate and raise awareness of how everyday individuals walk this Earth with an invisible disability.

Forty-five thousand individuals with Usher’s Syndrome are living in the United States, states The Coalition for Usher’s Syndrome Research. In reality, it is the leading cause of deaf-blindness. However, we have been given the opportunities to hear with hearing aids and cochlear implants, but none of us have effective treatments for blindness. We want to gain independence by driving and seeing picturesque destinations on our own, but we are limited to our vision loss. One of the first components of vision loss is night vision. Many struggle with the ability to see the beautiful, illuminating stars in the nighttime sky. Without treatment, the ability to see continues to decrease. Research for Usher’s Syndrome is underfunded by the National Institute of Health. According to The Coalition for Usher’s Syndrome Research, ALS affects 30,000 human beings and receives $47 million, but Usher’s Syndrome research receives only $3 million from the NIH. The Coalition for Usher’s
Syndrome Research hopes to receive more funding and find a cure someday in the near future.

Transitioning through the confusion and the denial, this Usher’s Syndrome diagnosis sparked a passion within me to continue and help the education and awareness of deaf-blindness. I want this thesis to deliver an insight to those who do not possess a disability and educate these individuals on becoming more aware of the disabled community, and what it is like to live in a world not completely handicap-accessible. Contrariwise, this can provide an opportunity for individuals who have a disability, visible or not, to feel a connection or an understanding of how we are all in this invisible community together, sharing similar struggles of everyday life.
Introduction

I believe things happen for a reason. Many of us do not know why; nonetheless, these things set us on a path. It is our destiny to fulfill our purpose during our lifetime. Some discover their purpose early in life, while others find their purpose later on. Occasionally human beings’ destinies are fulfilled after they have left this Earth, through their powerful work that continues to inspire for generations. Either way, these events that occur help us to contribute to society for the greater good. At least some point during our young lives, we dreamt of becoming doctors, firefighters, teachers, policemen, and so forth. We knew we wanted to do something that could help change lives, small or big.

I believe my disability is my purpose in life. Surely, I do not mean my disability alone, but how it sparked a passion within me. The lessons I have learned; the burdens I have endured; the strength I have built from my disability has shown me this is my calling. I want to take my experiences and help change the lives of others whom have a disability like mine.

The purpose of this thesis is to provide an opportunity for those who have a similar disability to feel a sense of community and relation; to understand they are not alone. Usher’s Syndrome is an invisible community; not many know of its existence. Even though all of our stories are different, this raw journal creates a connection between those of us with a disability, invisible or not. Also, this thesis provides insight to those who do not possess a disability, and will educate and raise awareness of what some individuals with disabilities endure.
“Sound checking…One, two, three… Is this too loud?” I shook my head no. “Okay then, we are going to get started. You are going to repeat the words I say back to me.” I shook my head up and down as I heard the lady enunciate.

“Airplane…Cat…Road…Apple…Brick…” I sat in a dark, foam padded, space cube with headphones and wires hooked to me. I looked around only to see myself in the room. Vaguely, I could see through the double paned glass into the other side seeing a lady with long, blonde, wavy hair in a white coat. Shortly after the repetitive words, she moved onto something else as equally exasperating. “Now, you are going to hear some beeping sounds. When you hear them, press the red button in your hand.” Vividly, I remember looking down into the palms of my hands and asking myself, “Why am I here? This only makes me frustrated. I know I can’t hear everything…” With each beep, the sound subsided.

I remember feeling vulnerable. Just stripped internally. I did not know why I was going through all of these appointments and doctor visits. Only at the age of five could I remember what my favorite Junie B. Jones book was and wanting to play outside with the neighborhood kids. However, this was the “normal” thing for me to experience. I wore these little, battery-powered radios on my ears, or so-called hearing aids. Little did I know, this hearing disability was connected to something more in the future. At this point in time, I only knew myself to be hard of hearing.

When I started Kindergarten at Abraham Lincoln Elementary School, every morning this bright, yellow school bus picked me up at the end of my drive way. Each morning, I walked onto the bus, said good morning to the bus driver, and passed by waving hello to the other students. As I sat down next to one of my morning bus ride buddies, I felt
comfortable. I felt a sense of community within the group of students on this bus. Looking around, I saw Billy, Whitney, Jessica, and Max laughing and signing to one another. Within this group of kids, I felt “normal.” These kids were just like me. Amy tapped on my shoulder and she signed, “Want to learn how to make a rainbow?” At this point in time, I thought it was the coolest thing ever. I told her, “Yes! Would there be a pot of gold at the end of it?” We both started laughing. She pulled out a pencil and notepad, began to write down the ingredients needed and the steps to making the perfect rainbow. Looking back, we did not let our disability define us as children. Was it the innocence we had? The naïve, youthful part of us?

Learning and realizing how individuals with disabilities were discriminated against and mistreated, it wiped clean the naïve child in me. After a couple of years of being comfortable with the other hearing impaired students in a small classroom, the faculty of the hearing impairment department felt that I should become more mainstream and integrated with other students in the school. Entering 2nd Grade, I was no longer with the other students who were like me. I sat in a pod with kids who could hear. I tried to sign to them, but no one understood my secret language. Luckily, I had my interpreter helping me through this transition. One day, I offered to share my crayons with a fellow student in my pod. We began to work on our coloring assignment, and then she proceeded to ask, “What is that? What are those things in your ears?” Immediately, I felt embarrassed. I turned bright red, and told her they were my hearing aids. She continued, “Why do you have those?” Internally, I felt myself becoming ashamed of my hearing aids. I knew I was different, but this confirmed that I was different from everyone else in the classroom. Quietly, I said, “I cannot hear as well, so I need them to help me hear better.”
Later on in the school year, we were playing out in the playground for recess. It was a brisk, cool day. Autumn was nearing to an end. The sun was shining. Students were running around playing tag and jumping rope. I was playing in the sandbox. Suddenly, this lanky boy with short, buzzed hair approached me. I recognized him from across the hall. He was in the third grade. For some reason, he looked like he was too old to be playing on this side of the playground. He began kicking sand at me. I told him to stop. He continued and then pushed me down into the sandbox. Every time I tried getting up, he shoved me back down. “You look so weird with those things in your ears...What are they? Why are you here? You’re not like us...” He continued to shout demeaning words and phrases at me about my disability. The bell could not have rung any sooner. He ran off and I was left in the sandbox with little dignity left. I stood up brushing off the sand, fighting back the tears of fear and embarrassment. Moments like this stay forever in your mind. It forced me to hate myself for having the stupid hearing aids. All I ever wanted to know was why? This crushed me. Situations like this continued to happen, verbally and physically. Constantly, I was reminded that I had a disability. I did not understand why the bullies were so persistent on making me feel like an outcast. Having my interpreter there did not help much, either; it only added to the unnecessary attention. Once I figured out a way to see the school nurse, Ms. Montgomery, I did everything I could do to keep seeing her. I would fake stomachaches or have a “cough.” Ms. Montgomery knew how to help me temporarily forget the bullying. I continued to pretend I was sick and not feeling well. I developed a hatred for the “normal” kids. Unfortunately, neither my parents nor my teachers knew this was happening. I was too embarrassed to say anything. All I wanted to do was go back to my friends who understood my secret language.
Luckily, I transferred to a private, Catholic grade school named St. Albert the Great. I used to be so mesmerized by the round, black stone church; the long hallway that stretched through the school, passing by students of elementary grades; and the tall ceilings in the gymnasium that sheltered us from the rainy days during recess. There was a family tradition within this school and church. My Mom’s family grew up there. My grandparents were proud to send my Mom and her siblings to this academic institution. I felt like I was becoming a part of this legacy. During this time, my cousins Shawn and Andrew were in attendance at St. Albert’s. Shawn was with the big kids in the school. Andrew, however, is only six months younger than me. When we both found out that we would be in the same school same class, we were ecstatic! Finally, someone I trust.

This is where I finished my remaining years before high school. This experience changed my perception of students who do not have disabilities. Not all people were mean or bullies against individuals with a hearing loss. Maybe I was mixed in with a bad batch of kids at the previous school? These students in my class became my family. There were only 18 of us, varying year after year. This gave me the opportunity to become independent in the classroom. No interpreter, no help from a teacher’s aid. Just me. I was able to feel “normal.” Equal to my peers. This experience taught me to become more individualized and take advantage of the positive aspects of my hearing disability. It taught me to become a better listener. I learned how to direct better focus on the individual speaking. I learned how to better process the information without the help of an interpreter. This opportunity gave me a second chance.

Entering high school, I realized it was a whole different game field. I will no longer be with the group of classmates I called my family for the previous six years. I had a sudden
urge to be discreet about my disability. Freshman year of high school, I had my hair long
and shaggy. Thankfully, the long, shaggy hair was trending within my generation. Through
this, I was able to blend in with everyone else avoiding the line of fire. I trusted very few
individuals, and I did not want to take any risks of experiencing the pain of the sandbox
encounter again.

One Thursday evening, I cut my shaggy, long hair short. It was out of impulse. I was
tired of the long, thick hair and wanted something new. A breath of fresh air. As the
hairstylist turned the chair around, I looked into the mirror. I realized you could see my
hearing aids. Seeing them wrapped around my ears, this exposed my insecurities. I thought
to myself, “What have I done? It’s so obvious now.” I knew there was no turning back. The
hair on the floor cannot be put back on my head.

The morning I went to school with a transformed haircut, I was standing at my
locker before first period, gathering my books for the morning. One of my close friends,
Kayla, walked up to me and said, “Heya!” Immediately, she recognized the new do and
complimented me. I felt good. Kayla proceeded to notice my Behind the Ear hearing aids.
She exclaimed, “I did not know you had hearing aids?! How come you never told me?!” “I
have a hard time hearing out of my right ear,” she laughed. After those words left her lips, I
realized that having a hearing disability is not a big deal. She wrapped her arm over my
shoulder and we walked down the hall to our first class. Nothing changed. She still treated
me like Brandon, not the teenage boy with hearing aids.

The day has come; I was turning 16 years old. Just like every teenager ready to peel
out onto the open road, I begged my mom to take me to the DMV to take my driver’s test so
I could get my license. My nerves were all tense, and I started getting butterflies in my
stomach when we approached the parking lot. My mom and I walked in, signed my name on the list, and patiently waited for my name to be called. “William...William Starkey,” proclaimed the attendant. I knew the time has come to prove my parallel parking skills. After the driver’s test, I was given an A-Okay to get my license. “Freedom at last!” I thought. Got my picture taken and went on my merry way. After I turned 16, I noticed my vision was not as strong as it used to be. Driving became a bit of a strain on my eyes, especially at night. I figured I might need glasses to drive. Then in general, I started noticing I could not see at night as well as I once could. One occasion, I drove over to a friend’s house for movie night. I shut off my car, grabbed my belongings, and walked up her driveway. It was dark, but it felt too dark. I could not see the moonlight shining onto the driveway. However, I continued to ignore the warning signs. At the time, I just figured it was the way I have always been able to see at night.

A couple months later, a close friend, Ben, called and asked if I wanted to go with him to a game night extravaganza. As any eager teenager to socialize, I said of course. This social gathering was in a basement at his friend’s house. The lights were all off, but some holiday lights were scantily stranded across the ceiling to set the ambiance. I locked onto my friend’s arm asking him to help me down the stairs. I could not seem to make shape of the floor. It looked unclear and hazy. “Maybe my eyes are just really tired?” I thought. “It has been a long day.” I noticed I struggled to recognize people’s faces in the poor lighting of the room. Suddenly, I felt uncomfortable. I felt like I was moving a lot slower than I should be naturally. I could not stand the anxiety any longer, and I had to leave the Apples to Apples contest. As I walked down the driveway to my car, I questioned, ”Do other people have a hard time seeing at night? Or even in dark spaces like that? Or is it just me?”
At the age of 19, I finally decided to mention something to my parents about the struggle I was having with my vision. Surely, I previously mentioned to them about getting glasses and correcting my vision with a prescription. However, I never truly expressed my concern for my visual strain at night. My parents were concerned and made an appointment with an Optometrist in my hometown. I met with Dr. Wiegman explaining my symptoms of flashes of light, floaters, and difficulty seeing at night or in any dim/poorly lit room. Dr. Wiegman performed the traditional eye exam, testing my vision through different lenses. Then, he proceeded to dilate my eyes. This I have never experienced before. My pupils were so large; they looked like the eyes of Hello Kitty. He shined a beam of light into my eyes and looked at my retinas. He noticed something that was peculiar. Dr. Wiegman suggested that I might have PVD, also known as Posterior Vitreous Detachment. He said, “Your vitreous sac might be pulling away from your retina, and that could be causing the flashes of light and floaters.” However, he was puzzled. “This does not typically occur in patients your age, though. It occurs mainly in elderly patients,” he added. Dr. Wiegman referred me to another local optometrist whom is a retina specialist that could provide me with a possible diagnosis.

Driving home from that appointment with Dr. Wiegman, I began to feel even more anxious and concerned. What if there was really something wrong? What if I was developing another condition? A few weeks later, I was in a bright, fluorescent white room with a robotic chair in the middle, surrounded by all of these gadgets and eye diagrams on the walls. Soon a frail, older man came into the room and introduced himself as Dr. Mann. He asked about my current symptoms and proceeded to notice I had a hearing disability. He continued to ask more detailed questions about my hearing loss than the reason why I
was in his office. I was muddled; I was there to see him for my eyes, not my ears. He dilated my eyes, and noticed my retinas were not normal in comparison to other patients my age. While he had this giant glass lens and bright beam of light shining into my eye, one of the phrases he muttered under his breath, “...bone spicules in both eyes,” to his medical assistant scribbling away. After he readjusted the robotic chair and turned the lights back on, he proposed the diagnosis of Retinitis Pigmentosa. It is a degenerative eye condition that leads to decreased vision and possible blindness. My heart stopped. I looked at my Mom and Dad with fear. What is going to happen from here on out? Then he asked if I have ever heard of Usher’s Syndrome? I said no. He suggested I get a genetic test performed to verify if I have Usher’s Syndrome or not. Dr. Mann explained, “Most patients who have a hearing loss similar to you in addition to Retinitis Pigmentosa (at your age) typically have Usher’s Syndrome.” At the end of the appointment he asked if I had any questions and I said no. I had hundreds of questions, but the shock and fear of knowing scared me into oblivion. Now I know why he was so curious about my hearing loss. Is this something rare?

I continued to meet with Dr. Mann. His medical staff performed different types of vision testing. Each time I went to his office, I felt like I was in the dark, foam padded space cube, five years old again. One appointment, I was sitting in a room with an egg shaped machine that had a small opening. I placed my head onto the chin rest, and watched the red dot in the middle. The nurse told me to sit still while they calibrated the machine. When the test began, I had to continue looking at the red dot in the middle in addition to looking for the green flashing light. I felt frustration building in my core, ready to burst at any moment. This was a visual field test testing my peripheral vision. I could hardly see the faint, flashing green light rotate across the inside of the egg shaped machine. I felt like I was failing the
test. I was struggling to see where the green flashing light was, and then it would appear into my visual field. In and out it would go. I realized when I could see the faint, green light; it was only a few spaces away from the red dot in the middle.

The last appointment I saw Dr. Mann, he entered the examination room and started talking to my Mom. My Father was in the waiting room since the examination rooms were too small to hold us all. I vaguely remember the initial conversation between my Mom and Dr. Mann. My surroundings became a blur. I felt with each visit to this retina specialist’s office, it was becoming more real of the possibility of having Usher’s Syndrome. I was lost in my own thoughts. Overwhelmed by the tests, the constant dilation of my eyes, and the poor vision test results. Suddenly, I come to; I hear scolding. Dr. Mann was criticizing my Mother and Father on their competency as parents. He demanded, “If you love your son, you will follow what I say…” I became angry. This man was scolding my parents for not following his specific directions? He instructed us to get my genetic testing done through the retina specialist at my university; however, it was not the facility Dr. Mann wanted the results through. After the fact, Dr. Mann insisted the testing to be done through Washington University, School of Medicine in St. Louis. Therefore, he was enraged, because he was not familiar with the formatting of the genetic testing results. The results we received were noninclusive, meaning the gene for Usher’s Syndrome they were searching for was not found. I had the urge to rebuke. Nothing came out. I was still numb. I looked to my Mother and saw her in tears. She was lost. Upset. Just like me. She believed we were doing the right thing. “We followed your instructions. My Mom and Dad are not incompetent parents, you narcissistic fool,” I thought.
We no longer scheduled appointments with Dr. Mann after that day. We were able to meet with a physician specialist at St. Louis Children’s Hospital. I had blood work done again to be mailed to a genetic testing lab in Iowa. I looked at each vile of blood being filled, thinking to myself, “This holds the answer.”

In July of 2011, I was diagnosed with Usher’s Syndrome, Type II A. All of these years, I thought I was only hearing impaired. I connected with my hearing disability more than anything. It was something I had since I was three years old. Now, I have a visual impairment in conjunction to my hearing disability? I was befuddled. The news came as a complete shock, and because of that shock, I was in denial. A part of me refused to accept that I was no longer just hearing impaired. There is something new I will have to make adjustments for in my lifetime. I was just a teenager. My life was just beginning. I was ready to start this new chapter of embracing the new opportunities of college. However, this put a temporary hold on my life, my aspirations.

Upon my diagnosis of Usher’s Syndrome, I had no idea what to expect or where to even begin. During the genetic counseling, she told me the basics. It is correlated with my hearing loss; therefore, it is a genetic condition that causes deaf-blindness. “Blindness?!” my subconscious screamed. I had no idea that this diagnosed genetic condition was the reason why I cannot see too well at night. I entered a denial phase. I did not even want to think about the idea of possibly going blind. I already handled the idea that I have a hearing loss, but losing my vision, too? “Why did this happen to me?” internally distraught. After the appointment, it was a quiet car ride home. I walked to my room and sat on my bed, staring into space. I sat there contemplating the world. I knew I had no direct answers. The physician said, “The symptoms vary for each individual person. One patient may have a
more severe case than you, or vice versa. All we can do right now is follow up and keep a record of your eyesight and the health of your retinas.” Thinking to myself, “All we can do right now is just sit and wait?” I did not want to accept the fact that one day; the world I see now will no longer visually exist.

One afternoon, I decided to Google what Usher’s Syndrome was. After I pressed enter, Pandora’s Box was opened. I read different articles and webpages only to find myself in tears. Upset. Broken. The information I read was not what I wanted to know. “Loss of night vision leading to loss of daytime vision, which could become tunnel vision,” I read. Several pictures of healthy retinas in comparison to retinas with Retinitis Pigmentosa lined the computer screen. “Pardon!” my subconscious knotted. This genetic disability is taking away another one of my five senses? The idea of losing my vision put me into an emotional lockdown. I refused to pity myself. I did not want to feel sorry for myself losing my vision. Just another weakness. It became a game of hurry up and wait. I wanted to accomplish my goals of earning a Bachelor’s, working in a field I enjoyed, starting and raising a family. “Will I be able to graduate within four years? Where will my vision be by the time I graduate? The accommodations I might need to help me to class, especially at nighttime. Working in the workplace will be different. I will need accommodations to help me complete tasks. Will this limit what I can do, and possibly make me incompetent? Furthermore, the thought of not seeing my children grow up into adults. Or will there be a chance they could have Usher’s as well?” whirled my brain. Revisiting these thoughts day by day drove me into a deep hole of despair. I did not know what to do. I understood there are individuals out there who are blind and are completely successful. However at this
point in time, I felt like my vision loss was a constant, daily reminder of my eyes slowly losing light, hope.

I was only 19 years old when I was diagnosed with Usher’s Syndrome, Type II A. I was experiencing college, figuring out what I wanted to do with my life, then all of the sudden my world stopped. Struggling to adjust and realize things really could be a lot worse was a feat. Some days I felt content, other days I felt lost. When I would come home from school, I could tell my vision was getting worse. Being out of an environment for a period of time and placing myself back into that setting put things into perspective. For example, my bedroom at night used to glow from the outdoor landscape lighting and streetlights, but I barely notice it anymore. I used to be able to see the silhouettes of the furniture and picture frames on the wall, but now it is just ambiguous and dark.

Another example, going to the movies is difficult. Before it used to not be a big deal at all. Go into the theatre, purchase your ticket and some popcorn, and then find a seat with your friends or family. Now, the theatre is too dim for me to find a seat without help. I cling to my Mom for guidance up the awkward, most nonconventional stairs to my seat. I struggle to remember: “Short, short, long, short long...short, short, long...” *stubs foot on off patterned step* I hate feeling dependent on someone else for help, especially in situations like this. The frustration I feel when I cannot just walk up the stairs in a movie theatre without tripping and struggling to see is hindering.

Most of the time, I avoid places like this. I think to myself, “Why put myself in those types of situations where I feel uncomfortable and my anxiety is high, because I have a nervous fear of tripping over something or not being able to see an object or person and collide into them.” However, I know I cannot always avoid places that are difficult for me to
see. If I know I am going somewhere that has poor lighting, I will logistically plan a route in my mind to help prevent the anxiety of the unknown. Most places I have been before, I memorize the layout. For example, if I have been to a restaurant previously, I mentally document if there are any steps, where they are located, and depending on how dimly lit the facility is, how many steps; where the restrooms are and which route is the most open so I can avoid any risk of running into a chair/table, etc. Everyday, I am constantly keeping record of different, little things that I think may be a risk for me if I do not see it.

In the summer of 2012, my Mom asked if I wanted to attend this conference for Usher’s Syndrome with her and my Dad. Initially, I was uneasy. I was not sure if I was ready for more information about Usher’s. My parents reassured me I have no pressure to go. It was up to me if I wanted to attend this event. My Mom informed me that there would be other individuals there with Usher’s. Later on, I decided that I would attend. I needed to be informed. “No matter how fearful I am of the information, I need to be educated,” I said to myself.

Early morning, I woke up, got dressed, and my parents, Aunt Yvonne, and I traveled to downtown St. Louis to the Chase Park Plaza. Turning the corner, I looked to see the Chase and whispered to myself, “Here we go...” Walking in, I felt the support of my parents and Aunt Yvonne. I noticed the accessibility changes made for the attendees here: bright, neon yellow tape stretched across each step; the lighting was brighter than normal.

As we approached the Check-In table, I received my initial, overwhelming shock. There was a couple, a man and woman in front of us, talking amongst themselves. I continued to look at the gentleman. He wore sunglasses and held his wife’s hand. In the other hand, he had a walking stick. I continued to look at him and saw his Behind the Ear
hearing aids. My heart started to race. I could feel the soreness of denial breaking down inside of me. My eyes began to water. I told myself, “No...you are not going to cry...you are not going to cry...” As I gathered my composure, I approached the table, received my materials and walked into the large conference room. I followed my Dad through the room, looked around and saw more people like the couple waiting in line. This time, I could not hold it in. I sat down at the round table, placed my head into my hands, and sobbed. This was a reality check for me. The realness within this room was too much to handle. “Am I going to be that man that was standing in line with his wife?” I thought. My Dad placed his hand on my back. He knew it was overwhelming for me. Just knowing that I had my parents and Aunt Yvonne there made me so grateful for their support. After a couple of moments, I controlled my breathing, dried my eyes, and pulled my head out of my hands.
Usher’s Syndrome: What is it?

Usher’s Syndrome is a rare, recessive genetic condition that causes deaf-blindness. According to the National Institutes of Health, a *syndrome* is a condition that possesses more than one feature or symptom. The major symptoms of Usher’s Syndrome are hearing loss and retinitis pigmentosa, or known as RP. Retinitis pigmentosa causes night-blindness and a loss of peripheral vision through the degeneration of the retina. The NIH states, “The retina is a light-sensitive tissue at the back of the eye and is crucial for vision. As retinitis pigmentosa progresses, the field of vision narrows, a condition known as ‘tunnel vision,’ until only central vision (the ability to see straight ahead) remains” (National Institutes of Health, 2011). Many people with Usher’s Syndrome also have severe balance problems.

![Figure 1](image)

*Figure 1: Photograph of the retina of a patient with Usher’s Syndrome (left) compared to a normal retina (right). The optic nerve (arrow) looks very pale, the vessels (stars) are very thin and there is characteristic pigment, called bone spicules (double arrows) (National Institutes of Health, 2011).*

Usher’s Syndrome consists of three clinical types: type 1, type 2, and type 3. According to the National Institutes of Health, types 1 and 2 are the most common in the United States. Together they account for approximately 90 to 95 percent of all cases of children who have Usher’s Syndrome (National Institutes of Health, 2011). Additionally,
approximately three to six percent of all children who are deaf and another three to six percent of children who are hard-of-hearing have Usher’s Syndrome. For example, in developed countries like the United States, about four newborns in every 100,000 births have Usher’s Syndrome. Statistically speaking, only 0.004 percent of a chance a child is born with Usher’s. This goes to show how rare this genetic condition is.

Since this genetic condition is inherited, both parents must have the gene to be passed onto their children. Scientifically speaking, genes are located in almost every single cell of the human body. Genes contain instructions that tell cells what to do. Each human being inherits two copies of each gene, one from each parent. Sometimes genes are altered, or mutated. “Mutated genes may cause cells to act differently than expected,” states the National Institutes of Health (National Institutes of Health, 2011). Through my research of Usher’s Syndrome, this condition is inherited as an “autosomal recessive trait.” The term “autosomal” means that the mutated gene is not located on either of the chromosomes that determine a person’s sex (National Institutes of Health, 2011). In other words, referring to the diagram on the left, both males and females can have the disorder and can pass the disorder along to a child. In terms of “recessive”, both parents must possess the Usher’s
Syndrome gene. If the child only receives one mutated gene of Usher’s Syndrome and a normal gene, the child is predicted to have normal vision and hearing. "Individuals with a mutation in a gene that can cause an autosomal recessive disorder are called carriers, because they ‘carry’ the gene with a mutation but show no symptoms of Usher’s Syndrome" (National Institutes of Health, 2011). Therefore, if both parents are carriers of the mutated genes for Usher’s Syndrome, it is a one out of four chance a child is born with Usher’s Syndrome each birth. Refer to figure 2 above.

According to the NIDCD, parents who have normal hearing and vision do not know if they are carriers of an Usher’s syndrome gene mutation. As of right now, it is not possible to determine if a family who does not have the history of Usher’s is a carrier. “NIDCD (National Institute on Deafness and Other Communication Disorders) scientists are hoping to change this, however, as they learn more about the genes responsible for Usher’s syndrome” (National Institute on Deafness and Other Communication Disorders, 2008).

Usher’s Syndrome consists of three clinical types: type 1, type 2, and type 3. Children with Type 1 Usher’s Syndrome are profoundly deaf at birth and have severe balance problems. Many of these individuals obtain little or no benefit from hearing aids. Parents should consult their physician and other hearing health professionals as early as possible to determine the best communication method for their child. Intervention should be introduced early, during the first few years of life, so that the child can take advantage of the unique window of time during which the brain is most receptive to learning language, whether spoken or signed. Also, if a child is diagnosed with Type 1 Usher’s Syndrome early on, before he or she loses the ability to see, that child is more likely to benefit from the full spectrum of intervention strategies that can help him or her participate more fully in life’s
activities. In addition, most children diagnosed with Type 1 have severe balance problems, children with this disorder are slow to sit without support and typically do not walk independently before they are 18 months of age. Also, these children usually begin to develop vision problems in early childhood almost always by the time they reach the age of 10. Visual problems for children with Type 1 most often begin with difficulty seeing at night, but tend to progress rapidly until the individual is completely blind (National Institute on Deafness and Other Communication Disorders, 2008).

The Coalition for Usher’s Syndrome Research discussed the second type. Type 2, is the diagnosis I received when I was 19 years old. This type consists of children born with moderate to severe hearing loss and normal balance. Although the severity of hearing loss varies, most of these children can benefit from hearing aids and communicate orally. The visual problems in Type 2 Usher’s Syndrome tend to progress more slowly than those in Type 1. Also, the onset of retinitis pigmentosa is often not apparent until the teenage years (The Coalition for Usher’s Syndrome Research, 2014).

The third type, Type 3, have normal hearing at birth. Although most children with the disorder have normal to near-normal balance, some may develop balance problems later on. Hearing and sight worsen over time, but the rate at which they decline can vary from person to person, even within the same family. A person with Type 3 Usher’s Syndrome may develop hearing loss by the teens, and he or she will usually require hearing aids by mid-to-late adulthood. Also, night blindness usually begins sometime during puberty, and blind spots develop by the late teens to early adulthood. Individuals with Type 3 Usher’s Syndrome is usually legally blind by mid-adulthood.
From all of these definitions and details about each type of Usher’s Syndrome and how individuals inherit this genetic condition, how is this syndrome diagnosed? Usher’s Syndrome affects balance, hearing, and vision. Therefore, the diagnosis of the disorder involves an evaluation of all three senses. Most individuals do not know they have this disorder until after symptoms appear. Through my personal experience, the evaluation of the eyes may include a visual field test to measure a person’s peripheral vision, an electroretinogram (ERG) to measure the electrical response of the eye’s light-sensitive cells, and a retinal examination to observe the retina and other structures in the back of the eye. For the hearing portion, a hearing (audiologic) evaluation measures how loud sounds at a range of frequencies need to be before a person can hear them. Lastly, balance is examined by an electronystagmogram (ENG), which measures involuntary eye movements that could signify a balance problem. Even though, most individuals do not know if they have Usher’s Syndrome until after symptoms show, an early diagnosis is still very important. The earlier that parents know if their child has Usher’s Syndrome, the sooner their child can begin special educational training programs to manage the loss of hearing and vision.

After the evaluation of balance, hearing, and vision, the only true confirmation of the diagnosis is through genetic testing. When I was visiting different doctors and audiologists, many of them recommended to have genetic testing performed. Through this, I was able to verify a concrete answer. Genetic testing for this syndrome is limited to 11 genetic loci (a segment of chromosome on which a certain gene is located) have been found to cause Usher’s Syndrome (Genetic Home Reference, 2007). Nine genes have been pinpointed that cause the disorder. Type 1 Usher’s Syndrome: MY07A, USH1C,
CDH23, PCHD15, SANS; Type 2 Usher’s Syndrome: USH2A, VLGR1, WHRN; Type 3 Usher’s Syndrome: USH3A. When my results came back, the genetic testing found the “USH2A” gene in my genetic sequence. Many individuals may have genetic testing performed, but receive no “yes” or “no” answer, because so many possible gene variations exist for this genetic condition. Therefore, genetic testing for this condition is not done on a widespread basis. Scientists are still attempting to find other genetic variations that cause Usher’s Syndrome.

For the time being, there is no cure for Usher’s Syndrome. The best treatment suggested by the Foundation Fighting for Blindness involves early identification so that educational programs can begin as soon as possible. The exact nature of these programs will depend on the severity of the hearing and vision loss as well as the age and abilities of the individual. Typically, treatment included hearing aids, assistive listening devices, cochlear implants, or other communication methods such as American Sign Language; orientation and mobility training; and communication services and independent living training that may include Braille instruction, low-vision services, or auditory training (Foundation Fighting Blindness, 2012).

Some ophthalmologists believe that a high does of vitamin A palmitate may slow, but not halt, the progression of retinitis pigmentosa. This belief stems from the results of a long-term clinical trial supported by the National Eye Institute and the Foundation for Fighting Blindness (National Institutes of Health, 2011). Based on these findings, the researchers recommend that most adult patients with the common forms of retinitis pigmentosa take a daily supplement of 15,000 IU (international units) of vitamin A in the palmitate form under the supervision of their eye care professional. (Patients with Usher’s Syndrome Type 1 did not take part in this study, high dose vitamin A is not
recommended for these patients.) For me, I have been taking two doses of an eye drop called, Dorzolamide (trade name Trusopt), every day twice a day. This ophthalmic solution is used to lower increased intraocular pressure in open-angle glaucoma and ocular hypertension (RxList, 2014). Even though I do not have glaucoma, the eye drops reduce the swelling of the retinas; therefore, prolonging vision life. By utilizing these eye drops, this helps to slow down the progression of the visual symptoms of Usher’s Syndrome. Some patients even receive steroid shots into their eyes for a more direct implementation to reduce the swelling. These are all research studies to see what could help patients with Usher’s Syndrome improve their vision life.

There is currently no cure for Usher’s Syndrome; however, there are researchers trying to identify all of the genes that cause Usher’s Syndrome and determine the function of those genes. This research will lead to improved genetic counseling and early diagnosis, and may eventually expand treatment options.
Transition from Denial to Acceptance

This newly “acquired” disability changed my perception on life. I developed to see the world in bright, vivid colors, as ironic as that sounds. From youth to adulthood, experiencing a disability put a heavy weight of responsibility on my shoulders. This accountability made me realize how I needed to figure out what is best for me. No one knows what is –internally- best for me. Surely physicians and audiologists may make suggestions and recommendations, but the individual living with the disability is myself.

This enlightenment process was a revelation. I mean this process was a process. It took several years before I became comfortable with my hearing disability, as will the adjustment to my newly “acquired” visual impairment. Being ashamed of my disability made me feel unequal to others. I had a weakness and could not hear as well as everyone else. Therefore, I was not “normal.”

As the clock counts down on the fourth quarter of my academic career at Indiana University, the appreciation of life I have established with the lessons I have learned puts me at awe. Surely with anyone, looking back you reflect on “If I knew then what I know now...” you would have been so much more wise. However, that is the part of the journey, learning to overcome obstacles and adversity to reach a full circle and understand your meaning, your purpose in life.

I have met several individuals who have inspired me to continue pursuing my ambitions, regardless of my hearing/visual impairment. Near my hometown in St. Louis, Missouri, The Megan Foundation helped me to realize life does not end when losing another one of your five senses. Meeting Megan, who was diagnosed with hearing loss at
the age of three, later to be diagnosed with the same Usher's diagnosis as me at the age of 22, was a moment I will never forget (The Megan Foundation, 2013). Internally, I felt we both connected instantaneously. This force of hope, pain, and uncertainty of what to come connected us. I looked into her eyes to see I was no longer alone. Megan made the transition from denial into acceptance much easier. This invisible community did not feel so invisible anymore.

During our lifetime, we find our destiny, our purpose. Through my journey, I discovered my calling. Overcoming many obstacles and adversities of my own, I realized my destiny is to educate and help raise awareness about Usher's Syndrome, as well as other disabilities. I knew I wanted to help people, one way or another. A goal of mine is to open a center providing resources for children and young adolescents who have a visual and/or hearing impairment like myself. Within this organization, resources like audiology, optometry, education, and psychological support for the individuals and their family and friends- transforming this invisible community to manifest into a network of individuals fulfilling their dreams and passions. Knowing people like Megan and how her foundation has impacted my life, I hope to change the lives of others, just like me.
Works Cited


